MISSOURI NEWBORN SCREENING

2015 Annual Report

Governor Eric Greitens
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Missouri Department of Health and Senior Services

Acknowledgments

The Missouri State Genetic Advisory Committee and its ancillary Newborn Screening Standing Committee, Sickle Cell Standing Committee, Cystic Fibrosis Standing Committee, Newborn Hearing Screening Standing Committee, and the Lysosomal Storage Disorder Task Force Committee play a vital role in supporting the activities of the Missouri Department of Health and Senior Services Newborn Screening Program.

The expertise the committees provide is complemented by department staff who are dedicated to helping Missouri children receive the best care available when diagnosed with one of the serious medical conditions detectable through screening tests.

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Missouri Department of Health and Senior Services
Division of Community and Public Health
Section for Healthy Families and Youth
Bureau of Genetics and Healthy Childhood
and
Missouri State Public Health Laboratory

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Screening Spotlight: Missouri's Newborn Screening Program Celebrates 50 Year Anniversary!

In 2015, Missouri celebrated 50 years of one of the most successful public health prevention and intervention endeavors since the smallpox and polio vaccination programs! Newborn screening (NBS) is a vital public health service that saves or improves the lives of 12,000 babies born in the United States each year. This amazing process of screening mass populations for a treatable genetic disease began in 1963 when Massachusetts passed a law to screen all babies born in their state for phenylketonuria (PKU). Later that same year, the states of Delaware and Oregon also passed legislation to begin statewide NBS. Each year after that, other states began passing newborn screening laws, and subsequently Missouri passed its NBS law in 1965. Now, every state in the U.S. has a NBS law and screens for more than 25 disorders.

In recognition of this exciting milestone in Missouri, the newborn screening program initiated a variety of awareness activities including an educational poster display that was set up at several state office buildings, a governor's proclamation, social media messages, and an informational pamphlet provided to genetic tertiary centers to distribute to their patients. These activities were carried out in order to celebrate Missouri's 50th year of newborn screening and also provide education regarding the impact and importance of the program.

NBS is the most efficient and most successful way to provide early detection for many rare but treatable disorders that need to be caught quickly after birth. Before birth, the baby's disorder is either compensated by the mother's physiology and/or has not had enough time to cause permanent damage to the baby. Here is a historical background on how NBS got started and how it has progressed:

- 1934: The discovery of PKU… Dr. Ivar Asbjörn Følling of Norway discovers that some of his mentally ill patients have high levels of phenylpyruvic acid in their urine, which shows a deficiency in an important enzyme to breakdown the amino acid phenylalanine. This deficiency is now known as Phenylketonuria (PKU).

- 1951: Discovery of Treatment for PKU… Dr. Horst Bickel, a German physician, discovers a treatment for PKU. He proves that a low phenylalanine diet can control the intellectual, developmental delay, and seizures caused by PKU.

- 1960: Dr. Robert Guthrie invents a test for PKU… Dr. Robert Guthrie, an American cancer researcher who had a niece with PKU, developed a simple and inexpensive bacterial inhibition assay which utilized a dried filter paper blood spot sample from a heel stick and could be used to screen for PKU in newborns and infants. Untreated PKU results in severe brain disability and the need for lifelong care of the child. For this achievement, Dr. Guthrie has been deemed the "Father of Newborn Screening".

- 1963: Newborn Screening Begins… After Dr. Guthrie published his findings regarding his very effective test and the results that early treatment can make, the state of Massachusetts passed the first NBS law to screen for PKU. Other states immediately followed suit throughout the coming years.

- 1965: Missouri passes its first NBS law… In the very beginning, a few large hospitals began conducting PKU testing on their newborns. Since PKU was so rare, individual hospitals were unable to maintain proficiency in its detection and eventually missed cases. Many babies from smaller hospitals were not even screened.

- 1967: Missouri State Health Laboratory takes over PKU screening… The decision was made to require that all PKU testing was to be conducted by the State Health Laboratory for all babies born in the state of Missouri, and so statewide PKU screening on all newborns was implemented at the Missouri State Public Health Laboratory. PKU has an incidence of around 1 in 15,000 infants.

- 1979: Congenital Hypothyroid Screening Begins… Treatment for congenital hypothyroidism (CH) is easy and inexpensive, and saves the affected newborns from severe brain and developmental disabilities. The screening methods for this disorder have improved tremendously over the years, and the positive predictive value of the screen has improved from 5% in the beginning to around 75% today. CH has an incidence of around 1 in 1,600 infants and is currently the most common disorder that we detect in the NBS laboratory. It is the only disorder on the NBS panel that is not genetically inherited.

- 1985: Galactosemia Screening Begins… Galactosemia presents the inability to breakdown galactose in milk and milk products. Although classical galactosemia only has an incidence of 1 in 50,000, without treatment, the disease is fatal within a few days or weeks after birth. That same year, the Missouri Genetics Advisory Committee was formed. Legislation required this committee to form and advise the Department regarding the NBS program's policies and panel of disorders.

- 1989: Sickle Cell Disease Screening Begins… Screening for sickle cell disease and other hemoglobinopathies was implemented as it was determined that prophylactic treatment with penicillin highly reduced the mortality rate in affected infants. These disorders have a combined incidence of 1 in 1,700 in Missouri. The testing method used also detects carriers of abnormal hemoglobins.

- 2001: Newborn Hearing Screening Begins… Screening for hearing deficiency was added to the required NBS tests; however this is conducted at the hospital with special hearing sensitivity equipment designed for babies. This is by far the most common newborn disorder, with an incidence of up to 1-3 in 1,000 infants. If hearing loss is not detected and managed early, it can impede speech, language, and cognitive development. Early detection allows special measures to be taken to keep children from falling behind in early developmental milestones, and also later on in school. This same year, the expanded newborn screening law passed and was signed by the Governor directing the department to add many other disorders to the NBS panel; in particular, amino acid, fatty acid, and organic acid disorders, along with congenital adrenal hyperplasia, cystic fibrosis, biotinidase deficiency, and G-6-PD deficiency. The testing would require two tandem mass spectrometers and several additional scientists, and could not begin until funding became available.

- 2002: Congenital Adrenal Hyperplasia Screening Begins… Screening for congenital adrenal hyperplasia (CAH) was implemented at the direction of the Missouri Genetics Advisory Committee and by the previously stated expanded screening law. CAH is a defect in the pathway leading to the biosynthesis of cortisol, and can result in ambiguous genitalia in females and salt-losing crisis in either males or females. Early detection and treatment is essential to prevent death in infants with salt-losing CAH. It has an incidence of about 1 in 13,000.

- 2005: Expanded Screening for Amino, Organic and Fatty Acid Disorder Screening Begins… With the addition of the Tandem Mass Spectrometry multiplex testing method to the NBS lab, an additional 41 metabolic disorders were added all at once, including the PKU testing that was currently conducted as a stand-alone fluorometric assay. The combined incidence of these disorders is around 1 in 2,000. Also during this year, the nationally Recommended Uniform Screening Panel (RUSP) was created by the American College of Medical Genetics (ACMG) and was endorsed by the March of Dimes.

- 2007: Cystic Fibrosis Screening Begins… Shortly before the move into the new State Public Health Laboratory, Missouri added cystic fibrosis (CF) screening to the NBS panel with the direction of the expanded screening law and the RUSP. CF is a genetic disorder characterized by severe lung damage and nutritional deficiencies. Early treatment can improve growth, improve lung function, reduce hospital stays, and add years to life. CF has an incidence of around 1 in 3,000.

- 2008: Biotinidase Deficiency Screening Begins… Biotinidase deficiency (BIOT) is a genetic disorder of impaired Biotin (vitamin B complex) usage and recycling. Children with profound BIOT, the more severe form of the condition, often have seizures, weak muscle tone (hypotonia), breathing problems, and delayed development. Treatment for this disorder merely requires biotin supplementation, and is easy and inexpensive. The incidence for profound BIOT is around 1 in 40,000, however, several milder forms of the disorder are found through routine NBS.

- 2012: Lysosomal Storage Disorder Screening Begins… Screening for Krabbe Disease began in August of 2012 in response to the Brady Alan Cunningham Act. The testing was temporarily contracted out to the New York State NBS Laboratory, which was the only other State laboratory in the U.S. screening for Krabbe. Missouri began the full population implementation phase for four other Lysosomal Storage Disorders (LSDs): Pompe, Gaucher, Fabry and Hurler Diseases in January of 2013. Missouri is the first State in the country to provide statewide screening and follow-up for these four LSDs, which are proving to have a combined incidence of greater than 1 in 2,000.

- 2013: Critical Congenital Heart Defect Screening Begins… Screening for critical congenital heart defects (CCHD) was added to the required NBS tests in January 2013. However, similar to the newborn hearing screen, this test is conducted at the hospital before discharge of the newborn. A routine and non-invasive testing method called pulse oximetry measures the oxygen saturation in the baby's blood from a finger or toe and can uncover heart defects that are not otherwise easily detected. Early intervention can prevent serious harm to the infant.

The lives of many Missouri infants have been saved or improved through NBS

Newborn Screening in Missouri

Disorder	Years of Screening	Babies Found
PKU	47	225*
Hypothyrodism	35	1,221*
Galactosemia	29	78
Hemoglobinopathies	25	1,174
CAH	12	52
Amino Acid (non-PKU)	9	32
Fatty Acid	9	137
Organic Acid	9	69
Cystic Fibrosis	7	184
Biotinidase Deficiency	6	67
LSD	19 months	66
		****Total = 3,305**

**The totals of babies found for the early years were estimated for this disorder using the average detection rates from the last 25 years.*

***This total does not include confirmed hearing deficiency and heart defects.*

Screening Spotlight: Parent Stories

"Our son's diagnosis of a rare metabolic condition called VLCADD at four days old was in a sense a blessing. However, it came as a shock with no family history of any health concerns and a baby that appeared perfectly healthy. While no parent wants to hear that their child has a life threatening disorder, by finding it as early as we did via his newborn screening, we were able to save his life. The early diagnosis allowed us to educate ourselves and care for our son in a way that not only saved his life, but has kept him a perfectly healthy, and happy child. I strongly urge all parents to take the opportunity to potentially save your child's life by taking part in newborn screenings."

A grateful mother in Missouri.

"Our son was born at the end of December 2007. We were so happy and excited that he was finally here. He was absolutely perfect!

After a week had passed, we received a phone call with unexpected news our son's newborn screen results detected that he had MCAD (medium chain acyl-coA dehydrogenase deficiency). Our world stopped. We were devastated and numb upon learning this news.

Today our son is doing great! Without the newborn screening, we do not know if we would be writing this to explain how grateful we are that the test, which revealed that our son has MCAD, was performed. We truly believe that the newborn screen saved our son's life! We are grateful that the screen was performed."

Parents of a son diagnosed with MCAD.

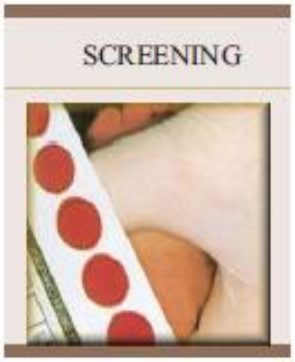
"On February 27, 2007 our daughter was born. She was a healthy, beautiful baby girl and we couldn't be happier that she was finally here. We took her home from the hospital and introduced her to our family and friends. Our family had a healthy new member with no complications, and we couldn't be happier.

Ten days after bringing our daughter home from the hospital things finally started getting back to normal in our household. Then we received a phone call on that day that the newborn screening test showed a positive result for PKU.

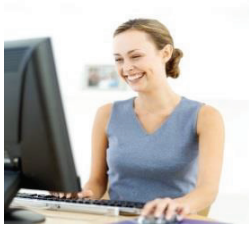
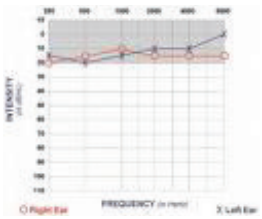
We are very thankful for the newborn screening test given to our child when she was born. Without the test we wouldn't have known anything was wrong with her until much later in her life when it would have been too late to help her. Because of the newborn screening test our daughter is reaching all her milestones and is a very intelligent little girl with nothing stopping her from reaching her full potential in life. It's all because of the newborn screening test. I'm sure we can speak for a lot of other families when we say that it was our daughter's savior to a higher quality of life."

Parents of a daughter diagnosed with PKU.

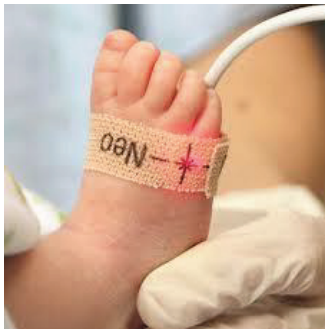
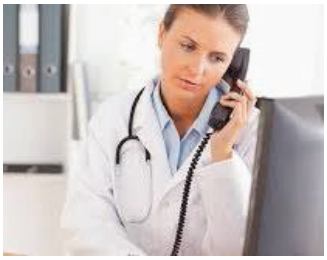
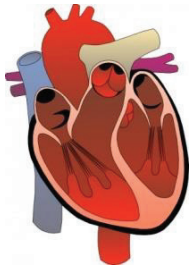
The Newborn Screening Process

1: TESTING	2: FOLLOW-UP	3: DIAGNOSIS/ INTERVENTION	4: TREATMENT & MANAGEMENT
• The baby's heel is pricked and a few drops of blood are collected on a filter paper 24 to 48 hours after birth.  • The dried blood spot specimen is shipped to the State Public Health Laboratory. • Specimen is tested for multiple conditions. 	• Positive screen results are reported by phone/ fax/letter from lab and follow-up staff to baby's physician. Results are also sent to the appropriate Genetic Tertiary Center in Missouri for follow-up.  • Specimen screening results are entered into data system. • Baby's physician or health care provider contacts baby's parents.  • Parents bring baby back in for evaluation and more testing at the genetic center.	• Depending on the screen result and the condition screened, repeat or confirmatory testing occurs at the genetic center.  • Parent education for signs/symptoms to watch for is conducted.  • Baby's physician consults with the specialist appropriate to the condition. 	• Once diagnosis is made, treatment begins. For some life-threatening conditions, treatment may occur prior to diagnosis - on the recommendation of a specialist.  • Parents receive treatment guidelines/ education. Team support services as appropriate, include: - Metabolic dietitian monitoring and consultation - Ongoing blood monitoring - Referral to early intervention services - Pulmonary/CF services - Pediatriac endocrine monitoring - Pediatric hematology monitoring - Genetic counseling and consideration of family testing - Other allied health services as needed

The Newborn Hearing Screening Process

1: SCREENING	2: FOLLOW-UP	3: EVALUATION	4: INTERVENTION
Baby is born. Hospital screens for hearing loss and checks for risk factors for late onset hearing loss prior to discharge.  Hospital submits results to the Missouri Department of Health and Senior Services (DHSS) via the Missouri Electronic Vital Records (MoEVR) system or on a paper form.  Manual data is entered into the Missouri Health Strategic Architectures and Information Cooperative (MOHSAIC) data system DHSS retrieves results from the MOHSAIC data system. 	Hospital reports results to parents and baby's physician.  DHSS sends letters to parents and physicians of newborns who did not pass or who missed the screening.  Parents return baby to hospital/health care provider 1-3 weeks after initial referral. 	Audiologist evaluates babies that don't pass a hearing screening by 3 months of age.  Audiologist reports evaluation results to DHSS.  Audiologist identifies risk factors and makes recommendations.  DHSS sends letter to families of children diagnosed with permanent hearing loss and refers to Missouri's Part C of the Individuals with Disabilities Education Act (IDEA) program, First Steps.	Babies diagnosed with permanent hearing loss enroll in First Steps (early intervention service) by 6 months of age. Babies receive services from the following as appropriate: Primary Care Physician, Otolaryngologist, Geneticist, and Ophthalmologist.  Baby may be a candidate for: hearing aids, cochlear implant, sign language instruction, or speech and language services. 

The Newborn Critical Congenital Heart Disease Screening Process

1: SCREENING	2: FOLLOW-UP	3: EVALUATION	4: INTERVENTION
• Baby is born.  • Hospital or midwife screens for critical congenital heart disease (CCHD) between 24 and 48 hours after birth or prior to discharge. • Screening should be in accordance with the American Academy of Pediatrics and American Heart Association guidelines.  • Screening should be done while baby is warm, calm, and awake. 	• If screening is normal, no further action is necessary. • If baby does not pass the screening, further evaluation will be necessary and the primary care provider should be contacted as soon as possible. 	• The baby's primary care provider will perform a thorough physical examination to rule out any non-cardiac issues that may have prevented baby from passing the CCHD screen.  • An echocardiogram may be done to look for a CCHD.  • The echocardiogram should be read by a pediatric cardiologist 	• Babies diagnosed with CCHDs will typically require surgical or catheter intervention within the first year of life.  • Parents will receive treatment guidelines and education.  • Babies may receive services from the following as appropriate: primary care provider, pediatric cardiologist, geneticist, nurse, nutritionist, pharmacist, social worker, and child life specialist. 

Newborn Screening Contact Information

Telephone Contacts:

Newborn Screening Laboratory main number	573-751-2662
Order newborn screening specimen forms	573-751-3334
Genetics and Healthy Childhood, for follow-up information	800-877-6246

Web Addresses:

Critical Congenital Heart Disease –
http://health.mo.gov/living/families/genetics/birthdefects/cchd.php

Newborn Screening Laboratory – http://health.mo.gov/lab/newborn/

Newborn Screening Program –
http://health.mo.gov/living/families/genetics/newbornscreening/index.php

Newborn Hearing Screening Program –
http://health.mo.gov/living/families/genetics/newbornhearing/index.php

Appendix 1: Disorders Confirmed for 2015 and Projected Incidence Rates

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Amino Acid Disorders	***14***	**1/6,000***
Arginemia		
Argininosuccinate acidemia	1	
Citrullinemia type I		
Citrullinemia type II		
Defects of biopterin cofactor biosynthesis		
Defects of biopterin cofactor regeneration		
Homocystinuria		
Hypermethioninemia		
Hyperphenylalaninemia		
Hyperphenylalaninemia, benign	1	
Maple syrup urine disease		
Maternal PKU		
Phenylketonuria (PKU)	10	
Tyrosinemia type I	1	
Tyrosinemia type II		
Tyrosinemia type III		
Methionine Adenosyltransferase I is not a disorder on the newborn screening panel but is found	1	
Biotinidase deficiency (BIOT)	***5***	**1/15,600***
Partial biotinidase deficiency	4	
Profound biotindase deficiency	1	
Congenital adrenal hyperplasia (CAH)	***4***	**1/19,500**
Congenital adrenal hyperplasia non salt water	0	
Congenital adrenal hyperplasia salt water	4	
Congenital primary hypothyroidism (CH)	***32***	**1/2,400**
Cystic fibrosis (CF)	***35***	**1/2,900**
Cystic fibrosis	27	
Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) or - Related Metabolic Syndrome (CRMS)	8	
Fatty Acid Oxidation Disorders	***18***	**1/4,590***
Carnitine acylcarnitine translocase deficiency		
Carnitine uptake deficiency		
Carnitine palmitoyl transferase deficiency I		
Carnitine palmitoyl transferase deficiency II		
Dienoyl-CoA reductase deficiency		

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Glutaric acidemia type II	1	
Long-chain hydroxyacyl-CoA dehydrogenase deficiency	2	
Maternal carnitine uptake deficiency		
Medium-chain acyl-CoA dehydrogenase deficiency	5	
Medium-chain ketoacyl-CoA thiolase deficiency		
Medium/Short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency		
Short-chain acyl-CoA dehydrogenase deficiency	4	
Trifunctional protein deficiency		
Very-long chain acyl-CoA dehydrogenase deficiency	6	
Galactosemia (GALT)	***17***	**1/4,300***
Classical galactosemia	1	**1/39,000****
Duarte galactosemia	16	
G-6-PD is not a disorder on the newborn screening panel but is discovered during testing for galactosemia	2	
Lysosomal Storage Disorders (LSD)	***46***	**1/1,730***
Fabry Disease	**27**	
Fabry	26	
Pseudodeficiency		
Unknown onset		
Genotype of unknown significance	1	
Gaucher Disease	**3**	
Gaucher type 1 (non-neuropathic)	2	
Pseudodeficiency		
Unknown onset		
Genotype of unknown significance	1	
Hurler Syndrome	**1**	
Attentuated		
Severe	1	
Pseudodeficiency		
Genotype of unknown significance		
Krabbe Disease	**1**	
Late infantile		
Later onset		
Pseudodeficiency		

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Unknown onset	1	
Genotype of unknown significance		
Pompe Disease	**14**	
Classical Infantile Onset	1	
Non-classical infantile onset		
Later onset	8	
Pseudodeficiency		
Unknown onset	1	
Genotype of unknown significance	4	
Organic Acid Disorders	***7***	**1/13,000***
2-Methyl-3-hydroxybutyric aciduria		
2-Methylbutyryl-CoA dehydrogenase deficiency		
3-Hydroxy 3-methylglutaric aciduria		
3-Methylcrotonyl-CoA carboxylase deficiency	1	
3-Methylglutaconic aciduria		
Beta ketothiolase		
Glutaric acidemia, type I		
Isobutyryl-CoA dehydrogenase deficiency		
Isovaleric acidemia		
Malonic acidemia		
Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)		
Methylmalonic acidemia (CBL, C,D)		
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)	1	
Multiple carboxylase deficiency		
Propionic acidemia	2	
Forminioglutamic acid (FIGLU) not a disorder on the newborn screening panel but is found	3	
Hemoglobinopathies	***50***	**1/1,500***
Sickle cell anemia disease (Hb S/S)	24	**1/3,000** Total population **1/400** African-American population
Sickle hemoglobin-C disease (FSC)	13	
Sickle beta zero thalassemia disease		
Sickle beta plus thalassemia disease (FSA)	5	
Sickle hemoglobin-D disease		
Sickle hemoglobin-E disease		
Sickle hemoglobin-O-Arab disease		
Sickle hemoglobin Lepore Boston disease		

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Sickle heredity persistence of fetal hemoglobin (HPFH) disorder		
Sickle "Unidentified"		
Homozygous-C disease (FC)	5	
Hemoglobin-C beta zero thalassemia disease		
Hemoglobin-C beta plus thalassemia disease		
Homozygous-E disorder (FE)	1	
Hemoglobin-E beta zero thalassemia disease		
Hemoglobin-E beta plus thalassemia disease		
Homozygous beta zero thalassemia disease		
Double heterozygous beta thalassemia disease		
Hemoglobin-H disease (Highly Elevated Barts)	2	
Other (FSX) compound heterozygous Hb S and G-Taipei		
Other Disease Condition		

*Combined incidence of all disorders in this category

**Incidence only for classical galactosemia

Appendix 2: Newborn Screening Laboratory Report - Samples Received 2015

	Newborn Specimens Received			Total Infant Specimens
	Initial	Repeat	Poor Quality	
Jan	6,164	1,226	155	7,545
Feb	5,614	1,102	147	6,863
Mar	6,511	1,246	163	7,920
Apr	6,039	1,178	108	7,325
May	5,867	1,135	98	7,100
Jun	6,587	1,223	124	7,934
Jul	6,687	1,284	132	8,103
Aug	6,521	1,157	109	7,787
Sep	6,644	1,346	158	8,148
Oct	6,238	1,385	156	7,779
Nov	5,937	1,125	128	7,190
Dec	6,285	1,402	170	7,857
Y.T.D.	**75,094 (82.02%)**	**14,809 (16.18%)**	**1,648 (1.80%)**	**91,551**

CALENDAR YEAR 2015
NEWBORN SCREENING LABORATORY REPORT
ABNORMAL RESULTS

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
BIO	Confirmed Profound	1	0	0	0	0	0	0	0	0	0	0	0	**1**
	Confirmed Partial	0	1	1	0	0	0	1	0	0	0	0	1	**4**
	High Risk	1	1	1	2	1	0	1	0	3	0	0	1	**11**
	Borderline Risk	2	3	4	9	10	4	1	3	18	5	1	8	**68**
CAH	Confirmed	0	2	1	0	0	1	0	0	0	0	0	0	**4**
	High Risk	5	3	5	6	6	2	4	2	4	2	1	2	**42**
	Borderline Risk	47	57	57	51	50	48	57	47	53	48	59	51	**625**
CF	Confirmed CF	3	3	1	6	1	1	2	2	1	3	1	3	**27**
	Confirmed CRMS	1	0	0	0	1	0	0	0	3	1	1	1	**8**
	Carriers Identified by NBS	12	14	11	10	11	8	9	7	8	10	11	14	**125**
	Referred	19	19	12	14	15	8	12	9	13	12	13	17	**163**
	Initial IRT	59	56	66	58	60	58	60	57	45	64	76	68	**727**
CH	Confirmed	3	1	5	3	2	3	4	2	1	4	3	1	**32**
	High Risk	4	2	8	3	2	4	4	3	6	4	3	2	**45**
	Borderline Risk	148	187	132	71	55	61	51	62	55	89	94	99	**1104**
GALT	Confirmed Classical	0	0	0	0	0	0	0	0	0	1	0	0	**1**
	Confirmed DG	1	0	1	2	2	1	1	2	2	0	0	4	**16**
	Confirmed G-6-PD	0	1	0	0	0	0	0	1	0	0	0	0	**2**
	High Risk	2	1	2	2	2	3	3	3	2	3	1	4	**28**
	Borderline Risk	2	3	2	4	5	4	6	4	6	3	4	4	**47**
AA	Confirmed	1	0	1	0	3	0	2	1	1	1	2	2	**14**
	Referred	3	1	2	1	3	0	3	2	3	2	3	3	**26**
	Low Risk	60	58	54	53	34	49	38	47	60	53	56	68	**630**
OA	Confirmed	0	0	0	0	0	2	1	1	0	2	0	1	**7**
	Referred	1	0	4	1	2	1	0	2	1	2	2	4	**20**
	Low Risk	39	48	39	37	50	30	39	21	34	44	51	36	**468**
FA	Confirmed	2	2	3	2	2	1	2	1	2	0	1	0	**18**
	Referred	6	5	4	5	5	5	5	3	6	0	4	2	**50**
	Low Risk	56	46	54	48	64	72	66	33	51	82	55	59	**686**
Hb	Sickle Cell Disease	3	1	3	3	2	2	6	2	5	3	5	6	**41**
	Other Hemoglobinopathies	0	1	1	0	2	0	3	0	0	0	1	1	**9**
	Traits Identified by NBS	135	118	136	132	125	146	151	129	137	142	127	133	**1611**
LSD	Confirmed Disorder	0	3	4	3	4	4	11	6	2	4	2	3	**46**
	Confirmed Carrier	0	1	1	3	3	0	2	4	3	3	0	2	**22**
	Confirmed Pseudo Def.	1	3	3	1	1	1	5	3	2	0	3	4	**27**
	High Risk	7	11	11	13	14	8	20	10	10	7	5	8	**124**
	Borderline Risk	0	0	0	0	0	0	0	36	24	15	25	15	**115**
SCID	Confirmed	0	0	0	0	0	0	0	0	0	0	0	0	**0**
	Referred	0	0	0	0	0	0	0	0	0	0	0	0	**0**
	Low Risk	0	0	0	0	0	0	0	0	0	0	0	0	**0**

Total Confirmed **230**

BIO = biotinidase deficiency
CF = cystic fibrosis
GAL = galactosemia
OA = organic acid
Hb = Hemoglobinopathies
CAH = congenital adrenal hyperplasia
CH = congenital hypothyroidism
AA = amino acid
FA = fatty acid
LSD = lysosomal storage disorder
SCID = Severe Combined Immunodeficiency

Appendix 4: Outcome Data – Newborn Screening Samples and Results

In 2015 there were 75,094 babies tested in the state newborn screening laboratory. There were 91,551 blood spot samples received in the laboratory. Samples received included:

Initial	Repeat	Poor Quality
75,094	14,809	1,648

In the process of screening newborns for 70 genetic and metabolic conditions, it is the newborn screening laboratory's role to assess the risk of any abnormal screening by evaluating the marker analytes and the levels that were detected. This risk assessment then dictates different levels of action and follow-up protocols. The 91,551 newborn screening samples received at the state newborn screening laboratory can be separated into two risk categories. The number/percentage of test results falling into these categories during 2015 were:

High Risk / Referred	Low Risk / Borderline Risk
559 (0.61%)	4,470 (4.9%)

High Risk / Referred – Results are immediately phoned and faxed to the physician of record and to the contracted genetic referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the facility that submitted the specimen and the physician of record.

Low Risk / Borderline Risk – Final laboratory results are mailed to the physician of record and submitting facility with a comment that a repeat newborn screen is necessary.

Two hundred and thirty (230) confirmed disorders were diagnosed from these abnormal newborn screen results during 2015.

Appendix 5: 2015 Poor Quality Samples Report

QUANTITY NOT SUFFICIENT: Quantity of blood on filter paper not sufficient for testing. Possible causes: removing filter paper before blood has completely filled circle; not allowing an ample size blood drop to form before applying to filter paper; inadequate heel stick procedure.	402
INCOMPLETE SATURATION: Uneven saturation; blood did not soak through the filter paper. Possible causes: removing filter paper before blood has completely filled circle or before blood has soaked through to opposite side; improper capillary tube application; allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood sample collection.	442
SAMPLE ABRADED: Filter paper scratched, torn or abraded. Possible causes: improper use of capillary tubes. To avoid damaging the filter paper fibers, do not allow the capillary tube to touch the filter paper. Actions such as "coloring in" the circle, repeated dabbing around the circle, or any technique that may scratch, compress, or indent the paper should not be used.	48
LAYERED CLOTTED OR SUPERSATURATED: Possible causes: touching the same circle on filter paper to blood drop several times; filling circle on both sides of filter paper; application of excess blood; clotted swirl marks from improper capillary application.	568
DILUTED, DISCOLORED OR CONTAMINATED: Possible causes: squeezing or milking of area surrounding the puncture site; allowing filter paper to come into contact with gloved or ungloved hands, or substances such as alcohol, formula, antiseptic solutions, water, hand lotion, powder, etc., either before or after blood sample collection; exposing blood spots to direct heat; allowing blood spots to come into contact with tabletop, etc. while drying the sample.	114
OLD SAMPLE: Sample greater than 15 days old when received at State Public Health Laboratory.	12
OTHER: Samples that did not elute properly and may be due to being either heated, improperly collected, or improperly dried.	8
NO BLOOD: Filter paper submitted without blood.	1
FILTER PAPER AND FORM BARCODES DO NOT MATCH: Barcode on filter paper does not match barcode on Newborn Screening Form. Collection forms contain barcodes on demographic, hearing, and filter paper portions. The barcodes may not be altered in any way. If incorrect baby is sampled, do not remove filter paper and	7

attach to a different demographic portion. If a sampling error occurs, the entire form needs to be voided and sample needs to be recollected on a new form. All barcodes must match laboratory copy, submitter copy, newborn hearing screen, and filter paper.	
MISSING, INCOMPLETE OR CONFLICTING PATIENT INFORMATION: Missing, incomplete or conflicting demographic information.	10
SERUM RINGS: Serum separated into clear rings around blood spot. Possible causes: card dried vertically (on side) instead of flat; squeezing excessively around puncture site; allowing filter paper to come in contact with alcohol, hand lotion, etc.	34
BLOOD ON OVERLAY COVER: Overlay cover came in contact with wet blood sample. Possible causes: sample is poor quality status because blood soaked from back of filter paper onto the gold colored backing of the form. The filter paper circles are designed to hold a specific quantity of blood. If the wet filter paper is allowed to come into contact with the paper backing of form, blood can be drawn out of filter making the quantative tests performed by the Newborn Screening Laboratory invalid. It is very important that the wet filter paper does not come into contact with any surface until completely dry.	2
Total Poor Quality Samples Received	1,648 (1.80%)

Appendix 6: Newborn Blood Spot Screening Hemoglobinopathy Report 2015

Specimens Received:

Initial:	75,094	(82.0%)
Repeat:	14,809	(16.2%)
Poor Quality:	1,648	(1.8%)
Total:	**91,551**	

Significant Results = 1,654					
Sickle Cell Disease		**Other Disease Conditions**		**Trait Conditions**	
FS	24	FE	1	FAS	1,071
FSA	5	Highly Elevated Barts	2	FSAINC	9
FSC	13	Other Disease Condition		FAC	325
FC	5			FCAINC	3
				FAE	37
				FAD	24
				FAX	122
				FASX	1
				FACX	0
				Slightly Elevated Barts	12
				Other Trait condition	0
Total	**47**	**Total**	**3**	**Total**	**1,604**

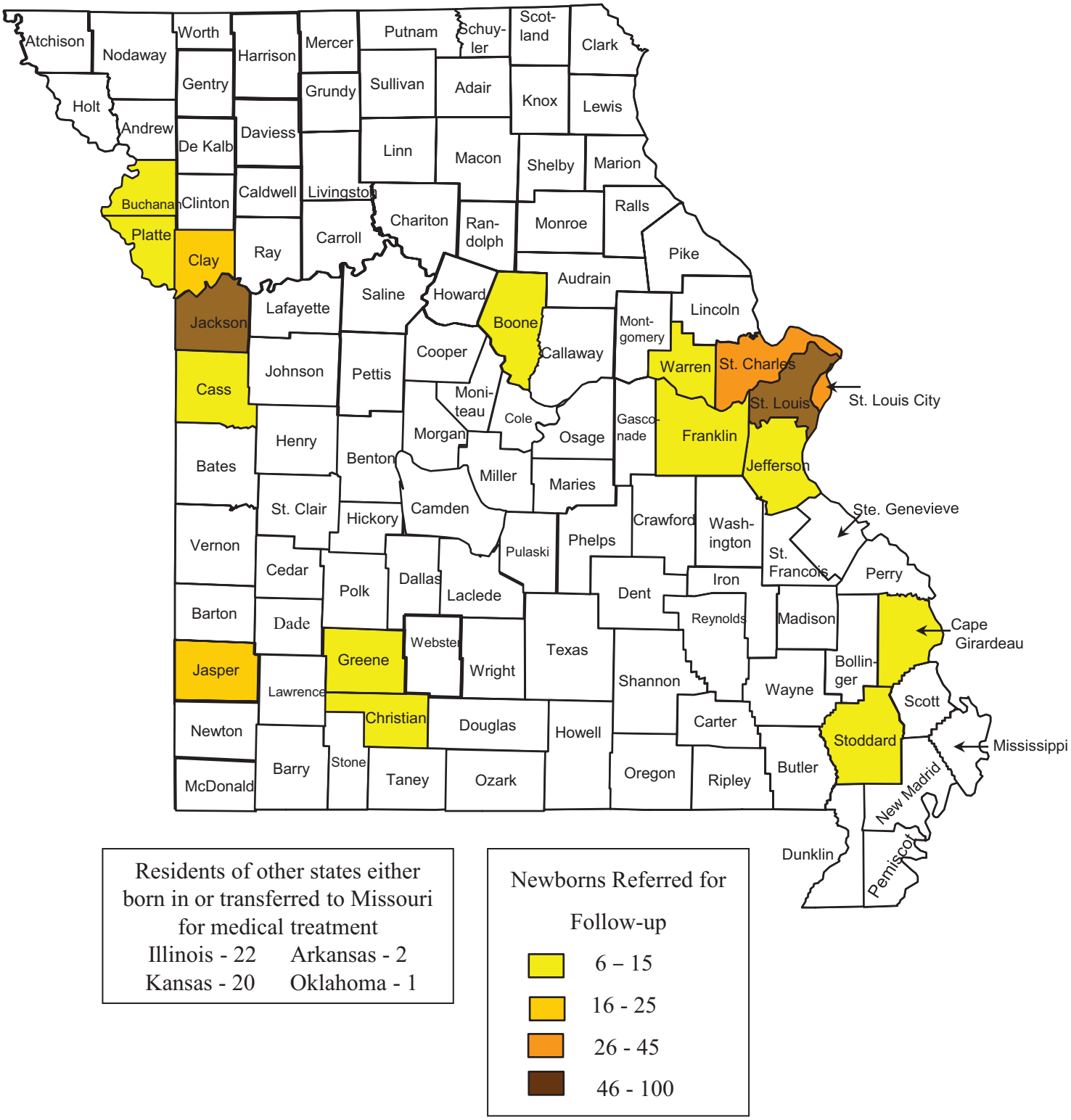
Appendix 7: Newborn Hearing Screening Data for 2015

2015 calendar year provisional data for Missouri shows:

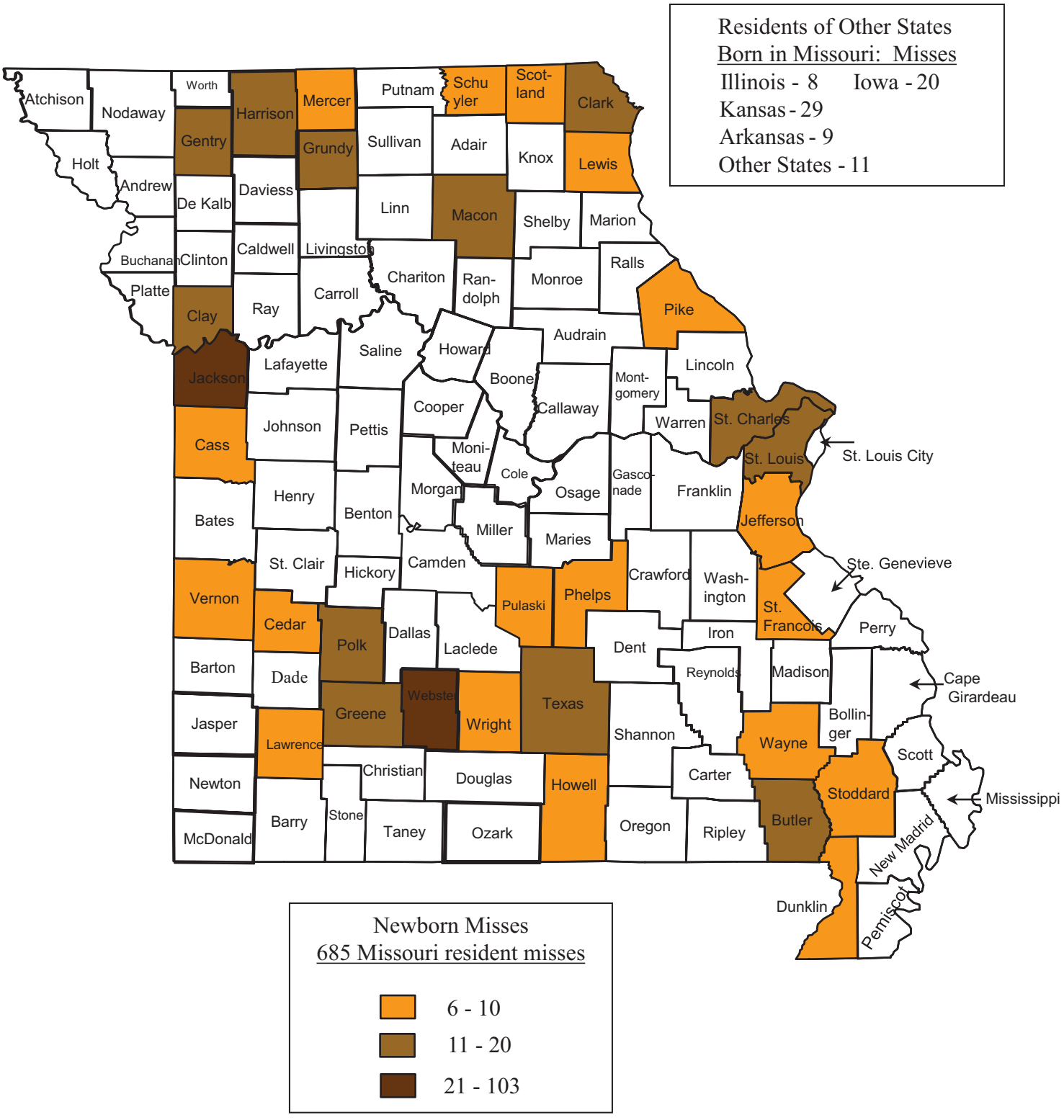
- 76,169 occurrent births (source: Department of Health and Senior Services Vital Records)
- 76,166 occurrent births (source: Missouri Health Strategic Architectures and Information Cooperative [MOHSAIC]*)
- 98.1 percent (74,731) of newborns were screened
- 97 percent (72,489) of infants were screened by 1 month of age
- 1.56 percent (1,172) of infants failed the final screening
- 77.8 percent (560) of the infants who failed their final screening and received an audiologic evaluation were evaluated and diagnosed by 3 months of age
- 121 infants were diagnosed with a permanent hearing loss
- 90 infants were enrolled in Missouri's Part C of the Individual with Disabilities Education Act (IDEA) program, First Steps
- 76.6 percent (69) of the infants enrolled in First Steps did so by 6 months of age

*The difference of 3 births between the occurrent birth count in the program data management system, the Missouri Health Strategic Architectures Information Collaborative (MOHSAIC), and the total occurrent births reported by Vital Records is the result of records that do not yet have an assigned Department Client Number (DCN) and records that are sealed. Records are not released from the Vital Records system to MOHSAIC until the DCN assignment is complete. Non-complete records are due to issues such as paternity and adoptions. Sealed birth records are neither displayed nor counted in MOHSAIC. This report is based upon MOHSAIC records.

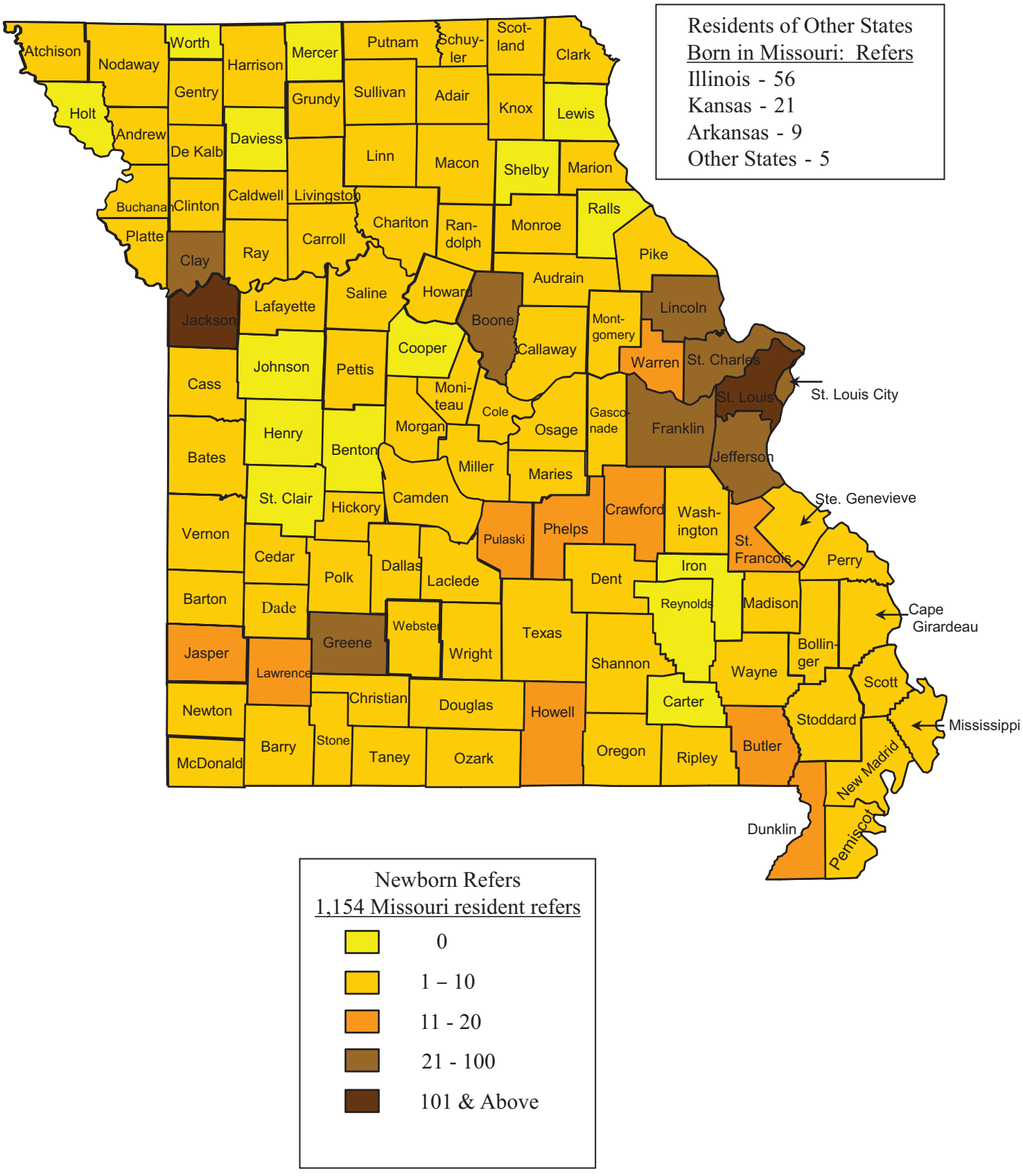
Appendix 8: Number of Newborns with Abnormal Newborn Blood Spot Screens Referred for Follow-up by County in 2015



Appendix 9: Number of Newborns that Missed a Hearing Screening by County in 2015



Appendix 10: Number of Newborns Referred After a Hearing Screen by County in 2015



Appendix 11: Newborn Screening Parent Satisfaction Surveys

A satisfaction survey of parents was conducted for families of babies having abnormal newborn screening results reported in 2015. There were 118 satisfaction surveys mailed and 17 were returned for a survey return rate of 14%. Key findings:

Newborn Screening Parent Satisfaction Survey			
	Very Satisfied	Satisfied	Not Satisfied
Staff explained my baby's condition in a way I could understand	94%		6%
Able to ask questions and discuss decisions about my baby's health care	94%	6%	
Offered reassurance and support	82%	12%	6%
The treatment staff was knowledgeable	88%	12%	
My questions and concerns were addressed in a timely manner	94%	6%	
The staff provided me with useful referrals and resources	94%	6%	
Received high quality care during my appointments	100%		

Reasons parents responded as not satisfied were because of a delay in testing additional family members, difficult to get a call back from the genetic counselor, and long wait time to have laboratory work done.

A satisfaction survey of parents and children receiving services provided by the hemoglobinopathy resource centers was conducted in 2015. There were 1,016 surveys mailed and 172 surveys returned for a survey rate of 17%. Key findings:

Hemoglobinopathy Resource Center Satisfaction Survey – Parent Response			
	Very Satisfied	Satisfied	Not Satisfied
Treated with respect	98%	1%	1%
Treatment staff was knowledgeable	98%	2%	
Questions/concerns addressed in a timely manner	97%	2%	1%
Staff provided useful referrals and resources	97%	3%	
Provided with the services needed	99%	1%	
Medical care/services received	88%	12%	
Received services or treatment without experiencing any problems	93%	5%	2%

Reasons parents responded as not satisfied were because the receptionist was not customer friendly and the wait time to see the doctor was more than an hour after scheduled appointment time.

Appendix 12: Newborn Hearing Screening Parent Satisfaction Survey

In February 2016, a 2015 satisfaction survey was mailed to parents of children born in Missouri who failed their initial newborn hearing screening between October 2015 and December 2015. There were 517 surveys mailed and 84 were returned for a survey return rate of 16%. The survey examined factors influencing the follow-up time between a failed newborn hearing screening and a repeat screening or an audiologic evaluation.

Key findings:

- 74% of the respondents reported that the birth hospital provided them with written information about the hearing screening prior to the hearing screening.
- 99% of the respondents reported that the birth hospital notified them of the screening result.
- 80% of the respondents reported that the hospital staff explained the importance of knowing whether a baby has a hearing loss early in life.

*Survey conducted every two years.

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